SMARCE1 gene

SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily e, member 1

Normal Function

The *SMARCE1* gene provides instructions for making a protein that forms one piece (subunit) of several different SWI/SNF protein complexes. SWI/SNF complexes regulate gene activity (expression) by a process known as chromatin remodeling. Chromatin is the network of DNA and protein that packages DNA into chromosomes. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. Chromatin remodeling is one way gene expression is regulated during development; when DNA is tightly packed, gene expression is lower than when DNA is loosely packed.

Through their ability to regulate gene activity, SWI/SNF complexes are involved in many processes, including repairing damaged DNA; copying (replicating) DNA; and controlling the growth, division, and maturation (differentiation) of cells.

The role of the SMARCE1 protein within the SWI/SNF complex is not completely understood.

Health Conditions Related to Genetic Changes

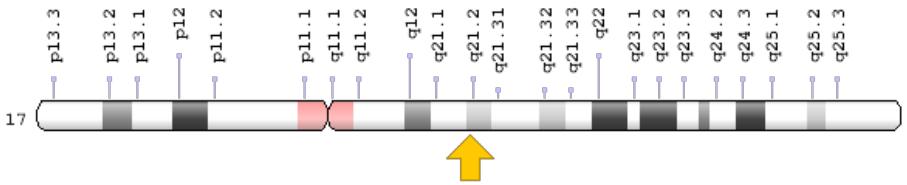
Coffin-Siris syndrome

At least one mutation in the *SMARCE1* gene causes Coffin-Siris syndrome, which is characterized by delayed development, abnormalities of the fifth (pinky) fingers or toes, and characteristic facial features that are described as coarse. This gene mutation changes a single protein building block (amino acid) in the SMARCE1 protein; the amino acid tyrosine at protein position 73 is replaced by the amino acid cysteine (written as Tyr73Cys). Although it is unclear how this change affects SWI/SNF complexes, researchers suggest that the mutation results in abnormal chromatin remodeling. Disturbance of this process alters the activity of many genes and disrupts several cellular processes, which could explain the diverse signs and symptoms of Coffin-Siris syndrome.

Chromosomal Location

Cytogenetic Location: 17q21.2, which is the long (q) arm of chromosome 17 at position 21.2

Molecular Location: base pairs 40,627,720 to 40,647,851 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BAF57
- BRG1-associated factor 57
- chromatin remodeling complex BRG1-associated factor 57
- SMCE1_HUMAN
- SWI/SNF-related matrix-associated actin-dependent regulator of chromatin e1
- SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily E member 1

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): ATP-Driven Chromatin Remodeling Machines Change Nucleosome Structure https://www.ncbi.nlm.nih.gov/books/NBK26834/#A644
- Molecular Biology of the Cell (fourth edition, 2002): Chromosomal DNA and Its Packaging in the Chromatin Fiber https://www.ncbi.nlm.nih.gov/books/NBK26834/

GeneReviews

- Coffin-Siris Syndrome https://www.ncbi.nlm.nih.gov/books/NBK131811

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SMARCE1%5BTIAB%5D%29+OR+%28%28BAF57%5BTIAB%5D%29+OR+%28BRG1-associated+factor+57%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D

OMIM

- SWI/SNF-RELATED, MATRIX-ASSOCIATED, ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY E, MEMBER 1
  http://omim.org/entry/603111

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SMARCE1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SMARCE1%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11109
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6605
- UniProt
  http://www.uniprot.org/uniprot/Q969G3

Sources for This Summary

- Hah N, Kolkman A, Ruhl DD, Pijnappel WW, Heck AJ, Timmers HT, Kraus WL. A role for BAF57 in cell cycle-dependent transcriptional regulation by the SWI/SNF chromatin remodeling complex. Cancer Res. 2010 Jun 1;70(11):4402-11. doi: 10.1158/0008-5472.CAN-09-2767. Epub 2010 May 11.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20460533
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2880201/
- OMIM: SWI/SNF-RELATED, MATRIX-ASSOCIATED, ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY E, MEMBER 1
  http://omim.org/entry/603111

- Santen GW, Kriek M, van Attikum H. SWI/SNF complex in disorder: SWItching from malignancies to intellectual disability. Epigenetics. 2012 Nov;7(11):1219-24. doi: 10.4161/epi.22299. Epub 2012 Sep 25. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23010866
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3499322/
- Tsurusaki Y, Okamoto N, Ohashi H, Kosho T, Imai Y, Hibi-Ko Y, Kaname T, Naritomi K, Kawame H, Wakui K, Fukushima Y, Homma T, Kato M, Hiraki Y, Yamagata T, Yano S, Mizuno S, Sakazume S, Ishii T, Nagai T, Shiina M, Ogata K, Ohta T, Niikawa N, Miyatake S, Okada I, Mizuguchi T, Doi H, Saitsu H, Miyake N, Matsumoto N. Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. Nat Genet. 2012 Mar 18;44(4):376-8. doi: 10.1038/ng.2219.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22426308

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/SMARCE1

Reviewed: May 2013
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services